Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Leukaemia Section

Short Communication

t(1;8)(p11-13;q24)

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Published in Atlas Database: April 2017
Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0108p11q24ID1454.html
Printable original version : http://documents.irevues.inist.fr/bitstream/handle/2042/68904/04-2017-t0108p11q24ID1454.pdf
DOI: 10.4267/2042/68904

Abstract

Review on t(1;8)(p11-13;q24), with data on clinics, and the genes involved.

KEYWORDS
chromosome 1; chromosome 8; MYC; Multiple myeloma

Clinics and pathology

Disease

Multiple myeloma (MM) mainly

Epidemiology

Multiple myeloma in 14 cases, found mainly in male patients (12 males and 1 female, 1 unknown) (Sawyer et al., 1998; Marzin et al., 2006; Wu et al., 2007; Gabrea et al., 2008; Sawyer et al., 2014.). Sporadic cases with lymphoid malignancies: CLL in a 64-years old male (Juliusson et al 1985) and T-cell acute lymphocytic leukemia (ALL) in a 1 year old boy (Raimondi et al 1993) (Table 1).

	Sex/Age	Disease	Karyotype
1	M/64	CLL	46,XY,add(9)(q34),del(11)(q22),+12,-13,-21,-22,+2mar/45,XY,+12,-21,-22/46,XY,t(1;8)(p12;q24)
2	M/1	T-ALL	46,XY,t(1;8)(p13;q24),t(11;17)(q23;q25)
3	M	MM	45-49,XY,t(1;8)(p11;q24),der(3)t(3;8)(p25;q?24),der(6)t(3;6)(q?21;q27),+der(7)t(7;?14)(q11;q11) t(1;14)(q11;q32),der(10)t(1;10)(q11;p15),-13,t(14;14)(q11;q32),+15,+18,+21 10-11.
4	M	MM	47,XY,-1,t(1;8)(p11;q24),t(2;3)(q31;q27),der(3)del(3)(p21)?dup(3)(q21q25),add(4)(p12),del(5) (q22q31),?del(6)(q21q22),inv(7)(p13q32),+der(11)t(11;15)(p15;q15),der(12)t(4;12)(p12;q24), del(13)(q12q14),der(16)t(1;16)(q21;q22),+der(19)t(1;19)(p13;q13),der(21)t(21;21)(q22;?)

5	?	MM	59,X?,t(1;8)(p1?;q24),der(2)t(2;11)(q3;?),-4,-8,-10,-12,del(12)(p?),-13,-14, +15,-16,der(16)t(9;16)(q11;q11),-18,+19,-20
6	M	MM	41-46,X,-Y,del(1)(p12p21),+i(1)(q10),t(1;8)(p12;q24),add(6)(q12),add(7)(p12),del(7)(p12),-8,add(12)(p13),del(12)(p11),-13,-14,+1-5mar
7	F	MM	39-41,-X,der(X)t(X;18)(q28;q11)ins(X;?)(q28;?),t(1;8)(p11-13;q24),add(2)(p25),dup(3)(q21q29),add(4)(q?31),t(4;14)(p16;q32),add(7)(p22),-8,der(9;17) (p10;q10),der(10)t(8;10)(q11;p13),-13,-14,add(15)(p11),der(15)t(9;15)(q12;p11),der(18)t(X;18),add(19)(q13),+20,der(21)t(1;21)(q23;p11)ins(21;?) (p11;?),+mar
8	M	MM	53,X,-Y,+1,t(1;8)(p13;q24)x2,+3,+6,+7,+8,+9,+11,t(12;22)(q13;q12),-13,+15,i(17)(q10),del(20)(q11q12),+21 19-20.
9	M	MM	55-56,Y,der(X)t(X;1)(q28;q12),t(1;8)(p13;q24),+der(1)t(1;8),+der(3)t(1;3)(q12;q29),+8,+9, +11, der(14)t(6;14)(p21;q32),+15,+17,+18+19,+20
10	M	MM	50,X,-X,+1,der(1;16)(q10;p10),+der(1;19)(q10;p10),t(1;8)(p11;q24),+4,+9,-13,?add(14)(q32), +15,+21, +21
11	M	MM	84,XXYY,i(1)(q10)x2,t(1;8)(p13;q24)x2,-2,-4,-4,-8,-9,i(9)(q10),t(9;12)(p22;q24)x2,-13,-13,-14,-14, add(14)(q32),-16,del(16)(q22)x2,+2mar
12	M	MM	53-56,X,-Y,t(1;8)(p13;q24),+i(5)(p10),+der(7)t(1;7)(q12;p21)add(1)(q?25),+9,+11,+15, +del(17)(p12),+18,+19,+20,+21,+mar/53-56,idem,-der(7),+del(7)(p12), del(12)(q13),+del(17)(q?22q?24), der(19)t(3;?;19)(p11;?;p13),der(19)t(12;?;19)(q11;?;p13)
13	M	MM	43-47,XY,+1,der(1;8)(q10;q10),t(1;8)(p11;q24),?del(2)(q?33),der(3)t(3;?8)(q?25;q?13),del(4)(q?32), -10,del(13)(q12q22),-16,add(16)(p13),del(17)(p12),der(19)t(1;19)(q12;p13)/44-45,idem,der(1)del(1)(p11p22)t(1;15)(q21;q22),+3,-der(3),del(6)(q21),der(15)t(1;15)(q21;q22),-der(19),+ider(19)t(1;19)(q12;p13)
14	M	MM	42,X,-Y,i(1)(q10),t(1;8)(p13;q24),add(2)(p?25),del(3)(p13p22),-4,t(5;14)(q11;q32),del(6)(q25), del(8)(p12),-10,-14,?del(18)(p11)
15	M	MM	81,XX,-Y,-Y,+1,+1,+del(1)(p13p32)x2,t(1;8)(p13;q24)x2,der(1;16)(q10;p10)x2,der(1;22)(q10;q10) x2,-2,-4,-10,-10,add(10)(q26),-13,-13,-14,-14,-17,?del(19)(q13)x2,add(20)(p13)x2/74-75,idem, der(1;15)(q10;q10)x2,-der(1;16),-der(1;22)x2,-3,-4,-5,-9,-12,-add(20),+22,+22
16	M	MM	61-65,XY,-X,t(1;8)(p13;q24),del(1)(p13),+del(1),der(4)t(1;4)(q12;q35),der(6)t(1;6)(q;q21),-10, add(12)(p13),-13,-14,-22

Abbreviations: M, male; F, female; CLL, chronic lymphocytic leukemia; ALL, acute lymphoblastic leukemia/lymphoblastic lymphoma; MM, multiple myeloma.
1. Juliusson et al., 1985; 2. Raimondi et al., 1993; 3-4. Sawyer et al., 1998; 5. Marzin et al., 2006; 6. Wu et al., 2007; 7-8. Gabrea et al., 2008; 9-16. Sawyer et al., 2014

Cytogenetics

Cytogenetics morphological

The most common breakpoints described on 1p were 1p13 (8 patients) and 1p11 (4 cases).

Additional anomalies

Found in a sideline in the CLL case (Juliusson et al., 1985) and in association with t(11;17)(q23;q25) in a T-ALL (Raimondi et al., 1993); highly complex karyotypes in MM showing mainly either pseudodiploid, hypodiploid or near-triploid/tertraploid karyotypes; associated with 14q32 rearrangements in 6 (Sawyer et al., 1998; Gabrea et al., 2008; Sawyer et al., 2014) and del(13q)/-13 was found in 10 MM patients (Sawyer et al., 1998; Marzin et al., 2006; Wu et al., 2007; Gabrea et al., 2008; Sawyer et al., 2014).

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

The chromosomal translocation t(1;8)(p11-p13;q24) is found mainly in multiple myeloma, indicating that it may play an important role in myeloma pathogenesis or progression of disease. While the translocation involve the MYC loci at 8q24, its rearrangement and/or overexpression was studied only in a few reported patients (Gabrea et al., 2008). Translocations involving MYC have been shown to be important in B-cell maturation or myeloma pathogenesis resulting in its increased expression and an aggressive disease phenotype. MYC translocations in MM are often complex with non-identified partner regions affecting non-IG partners (Walker et al., 2014). Therefore it is likely, that 1p11-13 is an additional partner loci not involving an IG gene causing dysregulation of MYC affecting proliferation, growth and apoptosis.

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This article should be referenced as such:

Zamecnikova A. t(1;8)(p11-13;q24). Atlas Genet Cytogenet Oncol Haematol. 2018; 22(5):213-215.